

Opis choroby *

Definicja

Orofaciodigital syndrome type 11 is an extremely rare, sporadic form of Orofaciodigital syndrome (OFDS; see this term) with only a few reported cases, and characterized by facial (blepharophimosis, bulbous nasal tip, broad nasal bridge, downslanting palpebral fissures and low set ears) and skeletal (post-axial polydactyly and fusion of vertebrae) malformations along with severe intellectual disability, deafness and congenital heart defects.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

OFD11

OFD11

Zespół ustno-twarzowo-palcowy, typ Gabrielli

Oral-facial-digital syndrome type 11

Oral-facial-digital syndrome, Gabrielli type

Orofaciodigital syndrome, Gabrielli type

Kod ORPHA

141000

Kod OMIM

612913

Kod ICD10

Q87.0

Kod ICD11

LD25.00

*Źródło

orphanet